The Technology and Innovation Unit of the National Institute of Health: A sequencing and bioinformatics core facility specializing in public health genomics

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The National Institute of Health (INSA) has a long tradition in investigating the molecular etiology of genetic and complex diseases. These activities greatly benefit from centralized sequencing services provided by the Technology and Innovation Unit (UTI). Its mission is to perform sequencing and genotyping assays in the framework of research, diagnosis and epidemiological surveillance, as well as to implement data analysis pipelines for the study of human gene variants. The equipment portfolio includes a *NextSeq 550*, a *MiSeq*, two *3500 AB Genetic Analyzers*, a *Fragment Analyzer* and a *7500 Real-time PCR system*. UTI provides results for average of 36.000 sequencing/fragment samples per year. The team has already performed >300 small genome, amplicon, gene panel (including clinical exome), *16S rRNA* gene and RNA/microRNA next-generation sequencing assays for INSA and for several Universities in the scope of scientific collaborations. Technical procedures are conducted under a quality control system that includes external quality assessment for next-generation sequencing/Sanger sequencing and ISO 15189 accreditation for Sanger sequencing. UTI plays a key role in public health genomics, providing state-of-the-art equipment, centralized resources, technical expertise and short response times. (This work was supported by Centre for Toxicogenomics and Human Health - UID/BIM/00009/2019 - and GenomePT project – POCI-01-0145-FEDER-022184)